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Supplementary Data

The Presenilin 1 P264L Mutation Presenting as non-Fluent/Agrammatic Primary Progressive Aphasia

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Supplementary Table 1
Previously reported clinical phenotypes of the *presenilin 1* P264L mutation. *Detailed clinical phenotype lacking but patients said to meet NINDS-ADRDA consensus criteria for a clinical diagnosis of Alzheimer's disease. –, absent or not described; AAO, average age of onset; FTLD, frontotemporal lobar degeneration; PPA, primary progressive aphasia

Publications	Origin	AAO (range)	No. Cases	Clinical diagnosis	Other features
Wasco et al., 1995 [1]	US	50	5	Alzheimer's disease	
Poorkaj et al., 1998 [2]	UK	41–45	5	Alzheimer's disease*	–
Campion et al., 1999 [3]	France	45–56	14	Alzheimer's disease*	Spastic paraparesis
Jacquemont et al., 2002 [4]	France	54	1	Atypical dementia with gait decline	Spastic paraparesis; Ideational apraxia; Impaired upgaze; seizures
Raux et al., 2005 [5]	France	47–51	4	Alzheimer's disease*	Spastic paraparesis
Dumanchin et al., 2006 [6]	France	55	4	Presenile dementia	Abnormal white matter on MRI
Martikainen et al., 2010 [7]	Finland	46–51	3	Alzheimer's disease ($n=1$) Dementia with Lewy bodies ($n=2$)	Spastic paraparesis Visual hallucinosis; extrapyramidal signs, delusions
Ishizuka et al., 2012 [8]	Japan	43–51	2	Alzheimer's disease ($n=1$) Atypical Alzheimer's/FTLD signs ($n=1$)	Anomia, comprehension deficits, personality change, disinhibition
Lohmann et al., 2012 [9]	Turkey	51	2	Atypical dementia – early episodic memory decline	Change in personality; frontotemporal atrophy on MRI
Current study	UK	45	1	Non-fluent/agrammatic PPA	Prominent early anxiety

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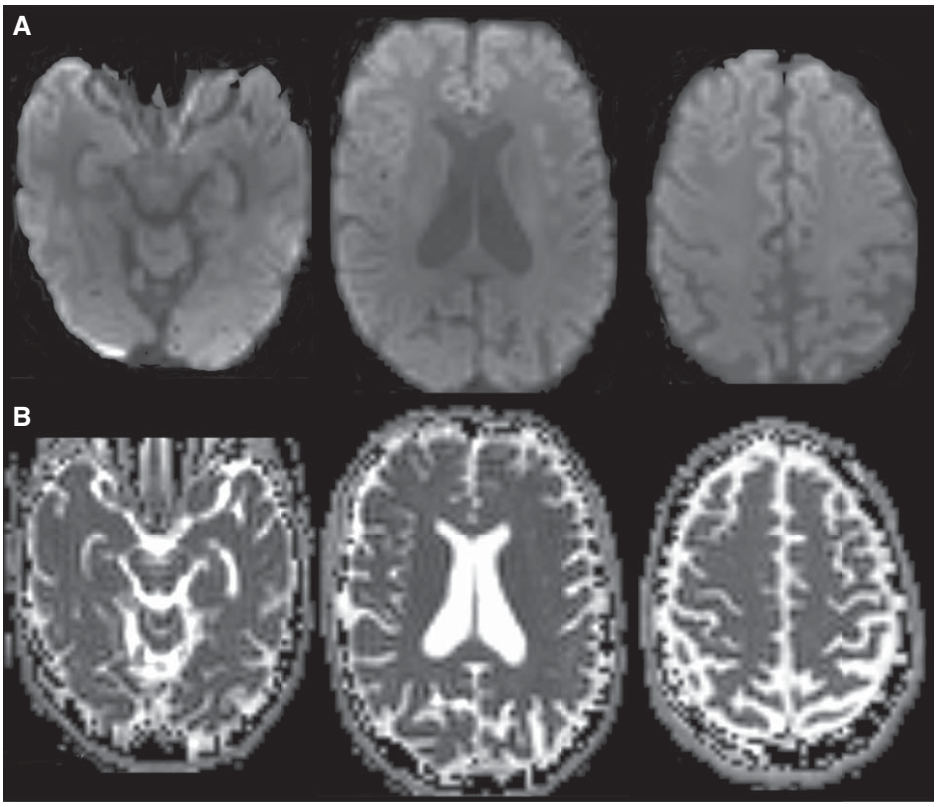

Supplementary Figure 1. Representative diffusion weighted imaging (A, b = 1000 s/mm2 images; B, Apparent diffusion co-efficient images) acquired axially.

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